Growing Dural Sinus Malformation with Associated Developmental Venous Anomaly, Multiple Cavernomas and Facial Venous Malformation in an Infant

An Associated Disease or a Disease Spectrum?

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Key words: dural sinus malformation, developmental venous anomaly, cavernoma, facial venous malformation

Summary

This is an unusual case report of an infant, who initially presented with a facial haemangioma and was later diagnosed to have a dural sinus malformation (DSM) involving the torcula. The DSM increased in size lateralising to the right transverse sinus at three months of age. Postnatal enlargement of the dural sinus has not been described before suggesting a delay in the maturation of the dural sinus which normally would occur antenatally.

There was a further association with a complex developmental venous anomaly (DVA) draining the right cerebral hemisphere into the deep cerebral vein and multiple cavernous malformations. The DVA was not clearly demonstrated at age one month but was more obvious at age three months. This would be the first reported case of DSM associated with a DVA. Increasing venous hypertension probably contributed to the poor opacification of the DVA on follow-up angiography at age six months and to the haemorrhagic changes within the cavernomas on magnetic resonance imaging (MRI).

The therapeutic goal was to correct venous hypertension by partially embolising the dural shunts to remodel the cerebral vasculature and preserve the patent sinus. The treatment strategy and possible link between the complex disease entities presented in this infant are discussed. Despite these attemps, the lesion continued to grow compressing the posterior fossa structures. The infant died at nine months of age.

Introduction

Dural sinus malformation in children generally occurs in the neonatal age group. It is one of the three types of dural arteriovenous shunts (DAVS) found in children; the other two are the infantile type DAVS and the adult type DAVS. DSM can be diagnosed in utero and are thought to correspond to an abnormal foetal development of the sinuses.

Anatomically, two types of DSM can be seen:

1. DSM involving the adjacent posterior sinuses have giant dural sinus pouches with very slow flow mural AV shunts. Partial thrombosis of the sinus may occur. The dural sinus pouch usually communicates with the other sinuses and drains normal cerebral veins. There are often restricted outlets due to thrombosis, occlusion or hypogenesis of one jugular bulb [1].

Enlargement (or ballooning) of the transverse sinus is a normal phase of sinus develop-

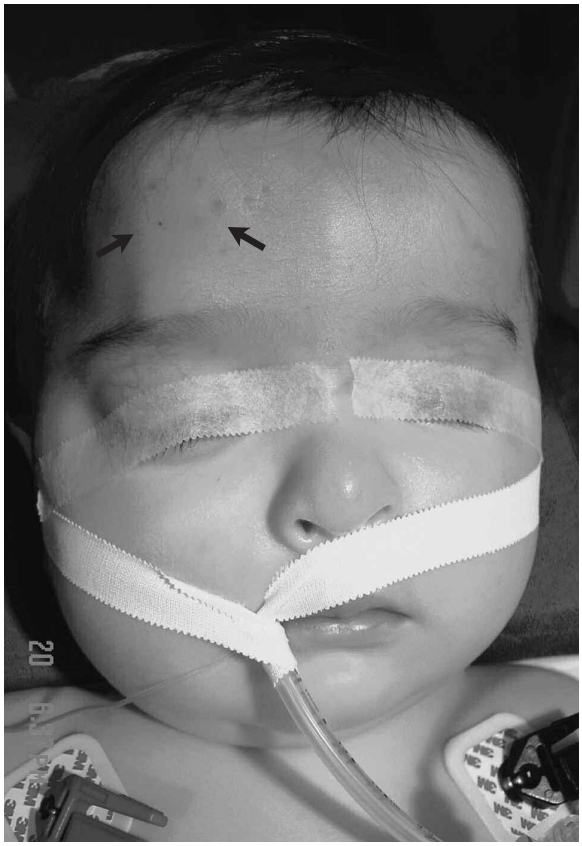

Figure 1 Photograph of the 6-month-old infant with right frontal cutaneous venous malformation (black arrow).

ment, during the fourth to fifth gestational months [2]. An unknown trigger has been postulated to cause the persistence or delay of the ballooning of the transverse and/or posterior part of the superior sagittal sinus (SSS). Important prognostic factors are the degree of lateralization (transverse, sigmoid sinus, jugular bulb) or mid-line location (torcula-SSS), magnitude of the dural pouch and thrombosis.

The evolution in midline located DSMs associated with slow flow multiple AV shunts within the wall of the dural pouch contribute to the venous congestion of the brain, initially without pial reflux. Spontaneous thrombosis of the pouch and outlets further compromise cerebral venous drainage and subsequently lead to venous infarction and intraparenchymatous haemorrhage.

As long as the venous outlets are patent, the clinical manifestations remain subacute and cerebral damage can be expected (being the natural evolution of the disease) taking the term melting brain syndrome or rapid loss of brain substance in midline lesions involving the torcula.

The treatment approach aims to preserve the dural pouch precluding a venous approach except in selected cases where it is possible to disconnect the brain drainage from the DSM without compromising it. The strategy is to preserve or remodel venous drainage to the brain and to occlude the mural AV shunts secondarily.

In lateralised malformations sparing the torcular, the capacity of the normal side to provide the necessary drainage for the brain is the most important prognostic sign. Similar favourable evolution is seen in the SSS locations away from the torcular [1].

2. Jugular bulb malformation with otherwise normal sinuses appears as a sigmoid sinus-jugular bulb "diaphragm" and is associated with a petromastoid-sigmoid sinus high flow AVF which is usually a single hole type.

The malformation has been postulated to correspond to a dysmaturation of the high jugular bulb [2], with occlusion of the sigmoid sinus, usually followed by the normal post-natal occlusion of the marginal sinus. The cerebral venous drainage is preserved and symptoms are mild in most cases. Prognosis is excellent with occlusion of the AVF, eventually with thrombosis of the sigmoid sinus distal to superior petrosal sinus opening.

Developmental venous anomalies [3] are non-pathological normal venous patterns. The deep and superficial types of DVAs constitute the extremes in the variability of the transcerebral venous system [4,6]. The actual embryological background is still uncertain, but it represents an extreme arrangement of the white matter venous drainage secondary to some abnormalities in venous development during embryogenesis. The deep variety of DVAs drain the normal subcortical territories of the superficial medullary veins into the deep venous collectors.

Complex developmental venous anomalies form a group of venous arrangements that include supratentorial and infratentorial anomalies on one or both sides of the midline. Complex patterns are rare but some have been encountered in association with facial lymphatic malformations and sinus pericranii [4,6].

Unlike lesions encountered in adults, most DVAs in children are not associated with cav-

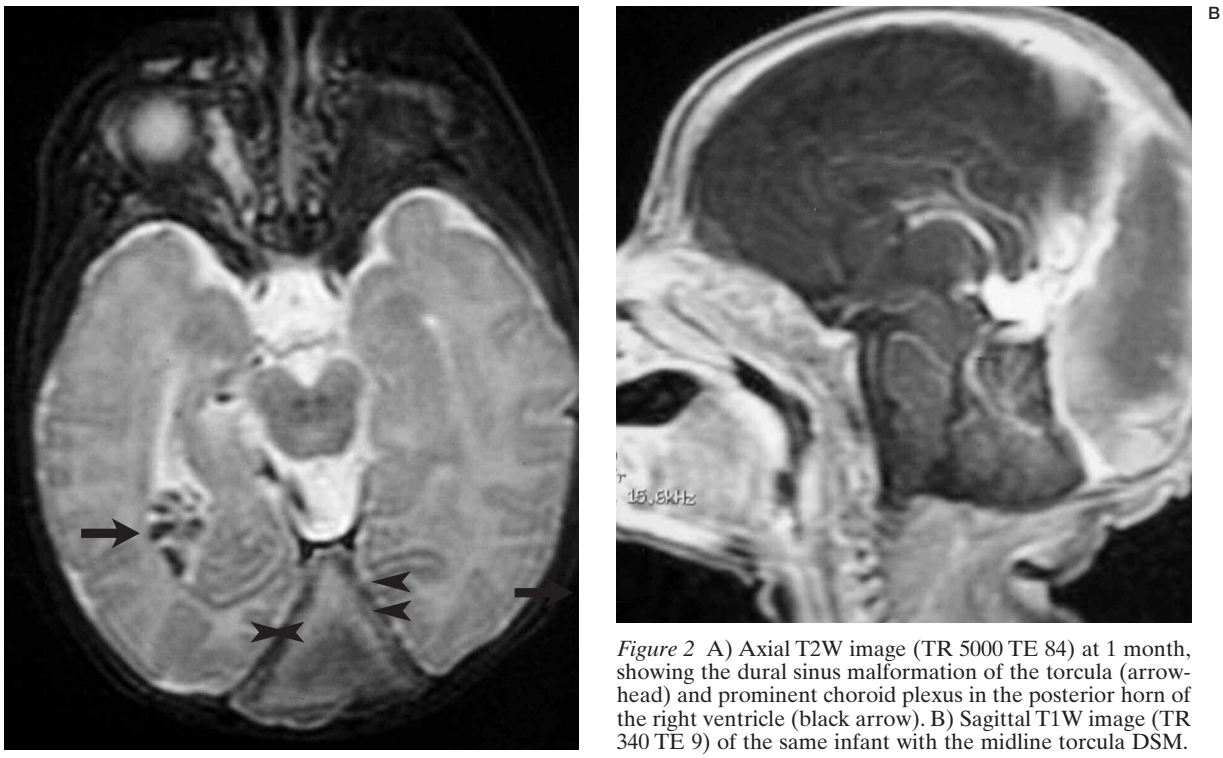

Figure 2 A) Axial T2W image (TR 5000 TE 84) at 1 month, showing the dural sinus malformation of the torcula (arrowhead) and prominent choroid plexus in the posterior horn of the right ventricle (black arrow). B) Sagittal T1W image (TR 340 TE 9) of the same infant with the midline torcula DSM.

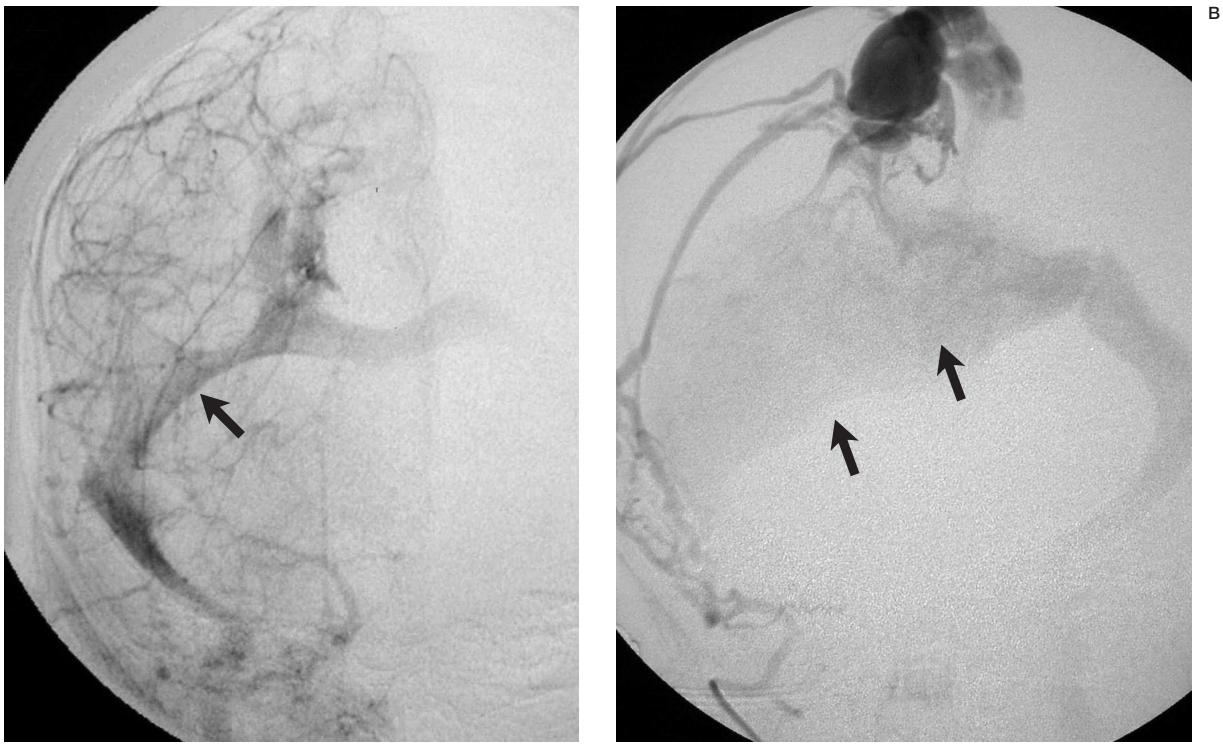

Figure 3 A) Venous phase of the RICA angiogram (frontal view) at 1 month, showing normal looking right transverse sinus (black arrow). B) Venous phase of the right internal maxillary angiogram (frontal view) at 3 months old demonstrating an enlarged right transverse sinus (black arrow).

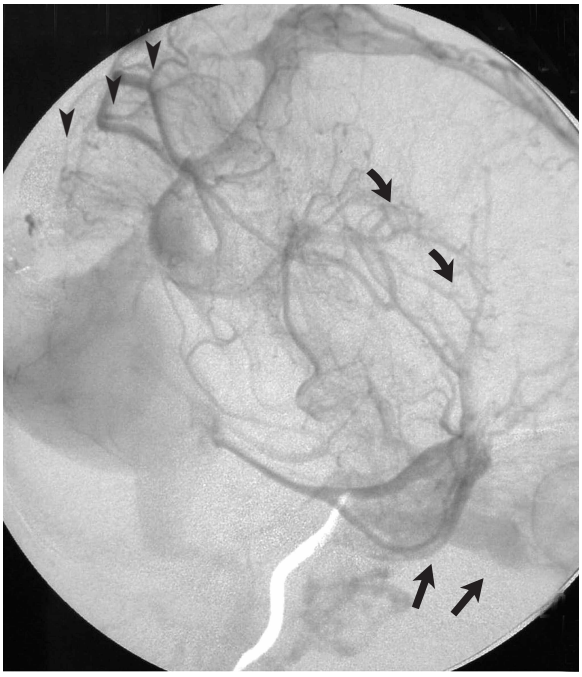

Figure 4 Venous phase of the right ICA angiogram (lateral view) at 6 months old demonstrating non-visualisation of the posterior SSS extending towards the torcula indicating thrombosis (arrowheads), abscence of cortical veins and dilated medullary veins (curved black arrow) converging into the deep cerebral vein with upstream drainage into the accessory falcine sinus and downstream drainage into the partially thrombosed torcula. Minimal cavernous capture demonstrated, draining into the dilated inferior opthalmic vein (black arrow).

ernous malformations. However, in cases of DVA with cavernous malformation, the association may be due to the peculiar haemodynamic conditions created by the DVA, which is capable of triggering an underlying defect and revealing as cavernoma over time. The secondary appearance of multiple cavernomas usually occurs in familial cavernous malformation and has been linked with a genetic site on chromosome 7q11.2-q21 [5,6].

The spectrum of cerebrofacial vascular syndromes has been well documented by several authors associating facial lesions with intracranial lesions, namely the rare disease cerebrofacial arteriovenous metameric syndromes (CAMS) a new name suggested by JJ Bhattacharya et Al, also known as Wyburn-Mason or Bonnet-Dechaume-Blanc Syndrome [1,10]. The typical feature of this syndrome is the associated high-flow arteriovenous malformation of the face with retinal and brain AVMs along the optic pathway.

In Sturge-Weber syndrome and DVAs, where thrombotic disease of the cerebral veins in the former and venous anomalies in the later entity have associated cerebral venous lesions with vascular (venous and lymphatic) malformations of the face [6-8].

In the complex neurocutaneous syndrome, haemangiomas of the head, neck and chest are frequently associated with persistence of the trigeminal artery, absence of the internal carotid artery or the vertebral artery (PHACE Syndrome) [7].

This case report describes an unusual presentation of a cerebrofacial vascular syndrome involving a dural sinus malformation (demonstrating postnatal enlargement) in association with a developmental venous anomaly, cavernous malformation and a facial venous malformation which has not been described before. The therapeutic challenge involved in treating this infant is discussed and the possible association between the complex disease entities is postulated.

Case Report

This six- month-old girl was born at term with a birth weight of 3.360 kg. She had multiple cutaneous naevi in the right arm, right anterior chest and right buttock. At one month old, she was noted to have a right frontal cutaneous hemangioma (figure 1), and an MRI done for the investigation of the facial lesion revealed a dural sinus malformation of the torcula (figure 2). The right choroid plexus in the posterior horn appeared more prominent than the contralateral side with an adjacent periventricular lesion consistent with cavernous malformation. She had no macrocrania or cardiovascular insufficiency at that time. The first angiogram performed at another institution revealed multiple dural AV shunts draining into the malformed torcula. However, both the transverse sinus, sigmoid sinus and internal jugular veins were patent (jugular bulbs were not yet formed). Cavernous capture did not occur and pial vein reflux was not demonstrated. The main arterial feeder was from the middle meningeal artery.

The infant was seen in Bicêtre when she was three months old and at that time, she had developed macrocrania and convergent strabismus of the right eye. There was no seizure or neurological deficit.

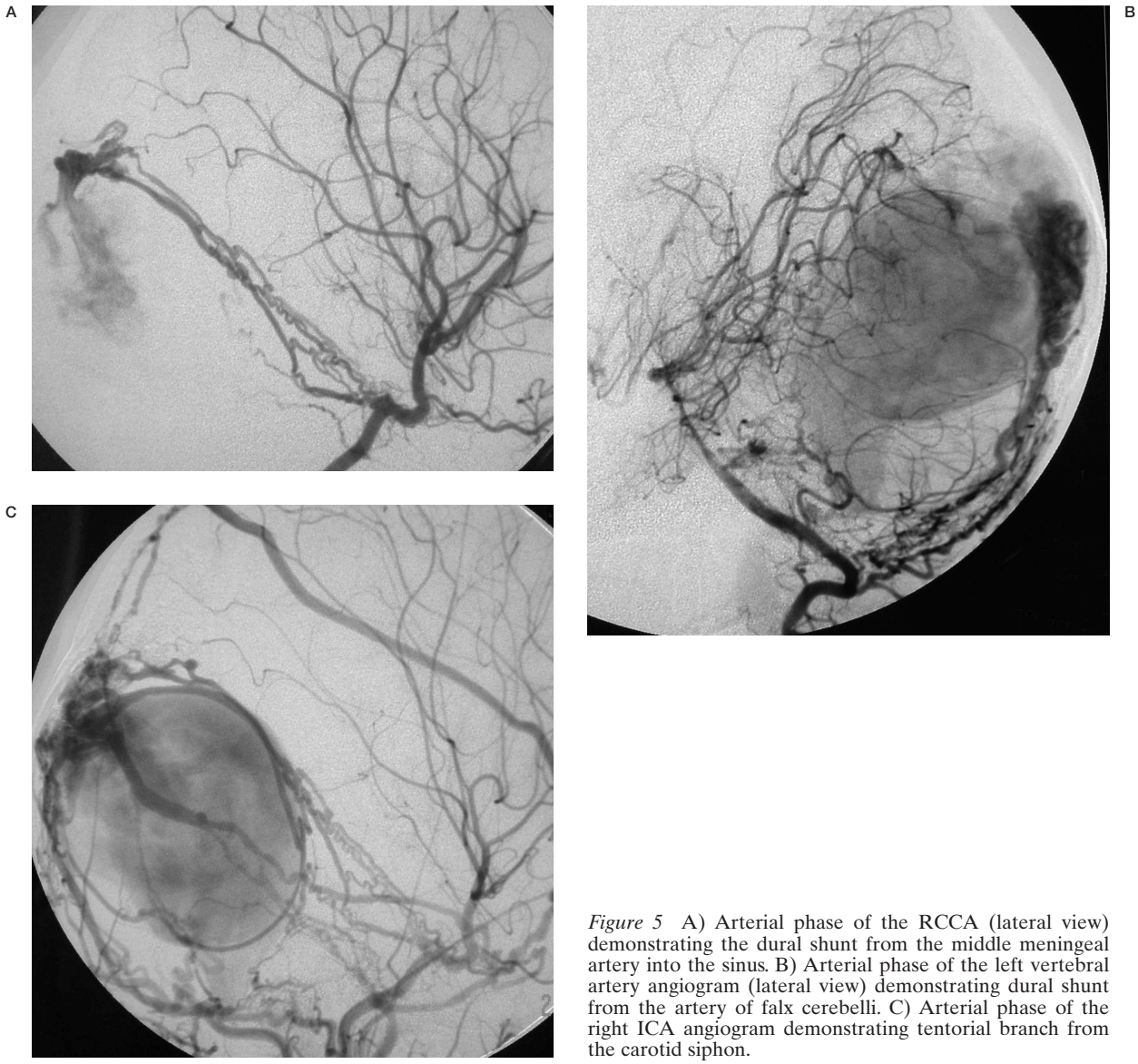

Figure 5 A) Arterial phase of the RCCA (lateral view) demonstrating the dural shunt from the middle meningeal artery into the sinus. B) Arterial phase of the left vertebral artery angiogram (lateral view) demonstrating dural shunt from the artery of falx cerebelli. C) Arterial phase of the right ICA angiogram demonstrating tentorial branch from the carotid siphon.

A second angiogram was done and revealed an enlarging sinus malformation involving the torcula and the right transverse sinus (figure 3). Partial thrombosis of the sagittal sinus (posteriorly) and the torcular had occurred. In the right ICA angiogram, a complex developmental venous anomaly drained the right cerebral hemisphere via the deep cerebral veins into the non-thrombotic portion of the sinus. Minimal cavernous capture was seen on the right side with extracranial drainage via a dilated inferior ophthalmic vein (figure 4).

This is unusual because normally the superior ophthalmic vein and/or the pterygoid plexus (low pressure draining system) brings relief to the remaining brain venous vasculature.

The mass effect of the enlarged inferior ophthalmic vein was probably the cause of the convergent strabismus in the right eye. Dural shunts were identified from the middle meningeal artery, the artery of the falx cerebelli and tentorial branch from the carotid siphon (figure 5).

The treatment strategy was to reduce venous hypertension by partially embolizing the dural shunts. Embolization of the shunt from the right middle meningeal artery was done with good resolution.

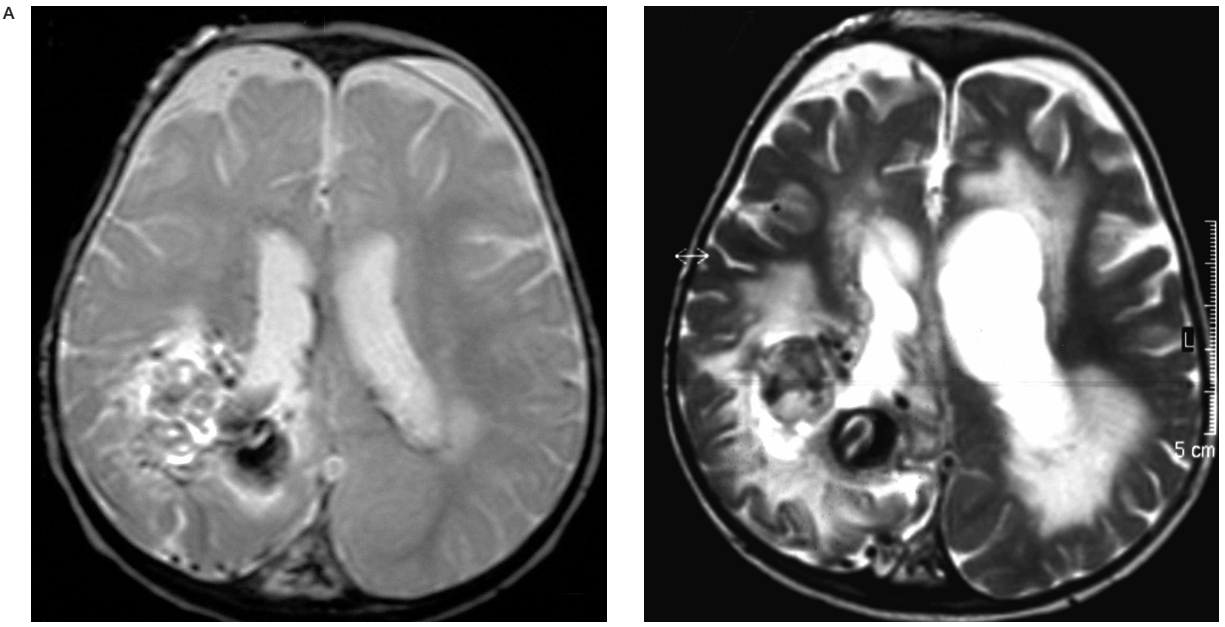

Figure 6 Axial T2W (TR 3965 TE 120) image showing multiple cavernomas with haemorrhagic changes in the right periventricular region.

At the age of six months, her psychomotor development was normal. A repeat angiogram still revealed multiple dural shunts. There was poor opacification of the right DVA and the deep cerebral veins with minimal cavernous capture. It was decided to embolize the shunts from the left middle meningeal artery to relieve the cerebral congestion. An MRI revealed multiple cavernomas with haemorrhagic changes in the right periventricular region posteriorly. There was some white matter oedema but not much mass effect (figure 6). On unenhanced axial CT brain scan, diffuse subcortical calcification was seen in the adjacent right parieto-occipital region which are signs of chronic venous congestion. The glue was seen as very dense material within the mural shunts in the SSS and torcula (figure 7).

The sinus as well as the cavernomas continued to enlarge. The child died at nine months of age. No autopsy was done.

Discussion

This child presented initially in the first month of life with a right facial haemangioma and an asymptomatic midline DSM involving the torcula and the posterior part of the superior sagittal sinus detected on MRI. Involvement of the torcula is not a good prognostic feature as it drains all the normal cerebral venous system at birth. Multiple dural shunts draining into the sinus malformation are characteristic of dural sinus malformation.

The clinical problem created by the midline malformation is the degree of reduction of venous outlets of the brain and the additional overload into the sinus by the shunts which contribute to the venous congestion of the brain. The endothelial property of the malformed portion of the dural sinuses seems altered, resulting in frequent spontaneous thrombosis. It is particularly rapid and extensive in midline giant lakes [6] which can lead to complete occlusion of the venous outlets causing venous infarction and sometimes coagulation factor consumption syndromes.

The child's situation gradually worsened angiographically with enlargement of the sinus malformation (at age three months) involving the right transverse sinus with partial thrombosis in the torcula and SSS. Post-natal enlargement of the sinus is highly unusual as it normally occurs in the antenatal period. Thus, one can postulate that the dural maturation in this child, was chronologically disorganized and delayed.

The angiogram also revealed a complex DVA draining normal brain of the right cerebral hemisphere into the deep cerebral veins.

These venous collectors converged towards the vein of galen (a high pressure system in this child) precluding the cortical system and the basal vein which normally would provide alternate collateral draining venous pathways to bypass the malformed torcula.

The DVA was not clearly demonstrated on retrospective review of the first angiogram done when the child was one month old. The cortical veins in the parietal region were not seen, but the cortical veins in the frontal region were present though smaller or hypoplastic compared to the left side. Can one postulate that the "compensatory venous drainage" which normally occurs in intrauterine life was delayed, similar to her dural sinus malformation which demonstrated postnatal enlargement?

The DVA being an extreme anatomic variation have reduced flexibility (adaptibility) and we can expect poor or no collateralization (dynamic system) compared to the normal venous disposition (static system). The increase in calibre in the medullary veins seen in this child, would be reflected as a form of embryonic or fetal adaptation of the DVA.

This would also be the first case to describe the association of a dural sinus malformation and a developmental venous anomaly. DVA can be associated with lymphatic malformations and other venous anomalies such as sinus pericranii, congenital vascular malformations (arterial, arteriovenous, venous) and acquired anomalies (ectasia) (table 1).

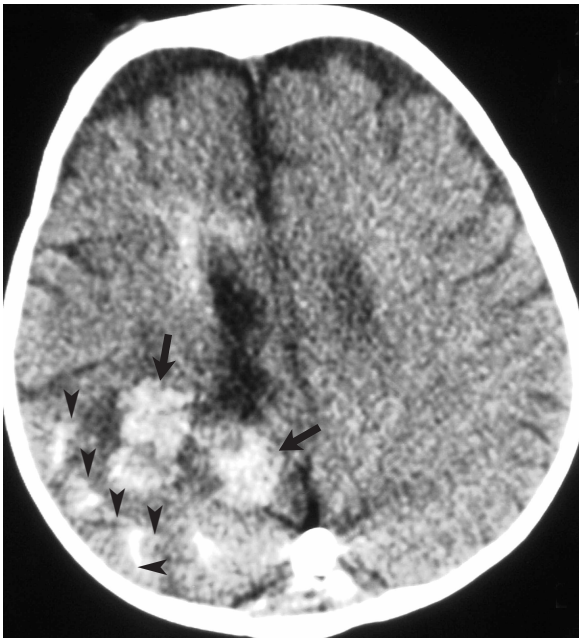

Figure 7 Unenhanced axial CT scan demonstrating diffuse subcortical calcification in the right parieto-occipital region (arrowheads), multiple cavernomas (black arrows) and dense glue material within the mural shunts of the SSS.

The facial haemangioma first noticed in this infant was actually a superficial venous malfor-

Table 1 **Reported combinations of disease entities in children and the present case has not been described before**

Reference	Dural AV shunts	Cavernous malformation	DVA	Facial lymp v.malf.	Sinus pericarinii	BRBN
(4,5)	+	+				
(10)		+	+			
(7)			+	+	+	
(3)			+	+		
(3)			+		+	
(3)				+	+	
To be published			+		+	+
Present case report	+(DSM)	+	+	+		

Note: BRBN – Blue rubber bleb nevus syndrome

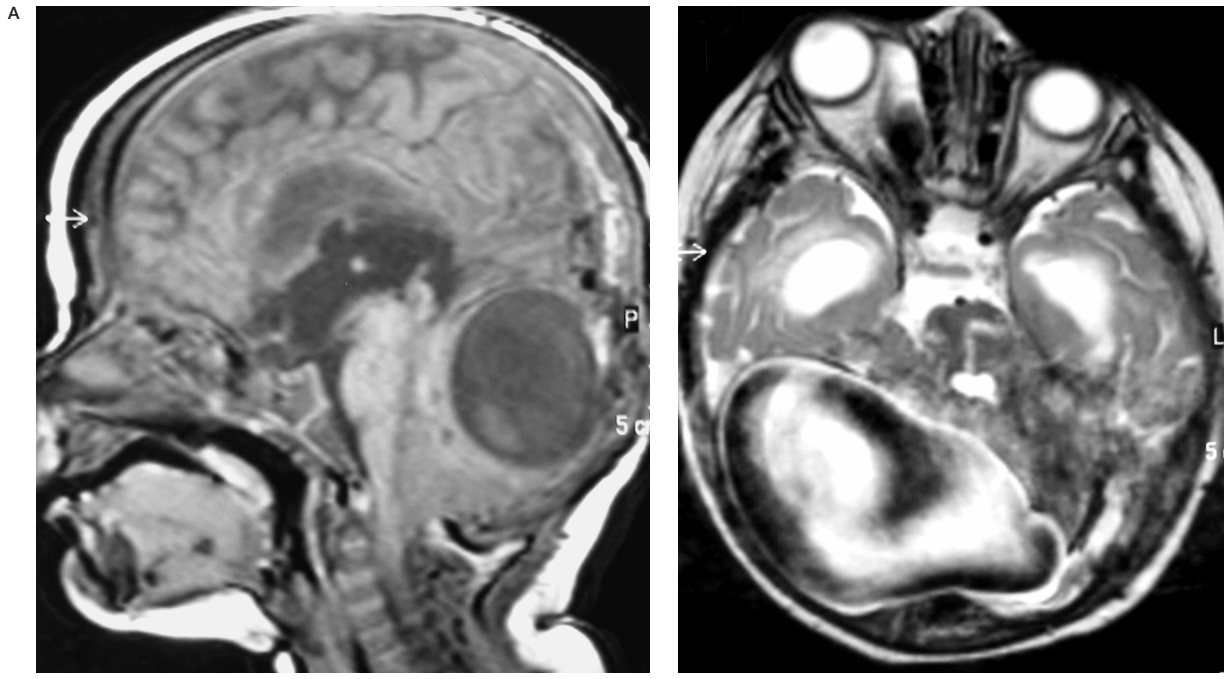

Figure 8 A) Sagittal and (B) axial T2W (TR 4877 TE 110) images at 9 months of age, showing the size of the torcula compared with figure 2A and 2B.

mation under bluish skin enhanced by Valsalva maneuvre (typically when the baby cries) and is more likely linked to the DVA rather than to the DSM.

Her condition was further complicated by multiple cavernous malformations which were initially single and silent in the earlier MRI (first month). The early appearance of the cavernomas is unusual even in the presence of coexisting DVA. One other case report [5] documented multiple cavernomas in an infant initially presenting with multiple AV shunts: dural and then pial AVM (table 1). The genesis of the cavernomas remains unclear.

With the concept of pathogenesis and natural history in mind, the treatment principle was to correct venous hypertension by partially embolising the dural shunts and preserving the patent sinus. Her first embolisation was done at three months when she presented with macrocrania, a sign of water retention.

The hydric system of the neonate is still immature, which results in the venous system being responsible for both venous drainage and the intrinsic and extrinsic cerebral water dynamics. Cerebral congestion will compromise venous and water drainage manifesting as macrocrania in this infant.

The DVA draining the right cerebral hemisphere was not well opacified with poor cavernous capture in the most recent angiogram three months after the first embolization. This poor prognostic sign suggests that there is a restriction of cranial venous outlet thus producing retrograde congestion. The problem of venous hypertension was exacerbated by the late opening of the cavernous sinus which could have provided relief to the remaining cerebral venous system. A direct relationship between the demonstrated reflux and cerebral damage will occur.

It was decided to proceed with the second embolization to reduce venous hypertension. Two shunts form the left middle meningeal artery were embolized intra-arterially using a mixture of NBCA and lipiodol. An MRI post embolization revealed multiple cavernomas which had bled in the right periventricular region. Fortunately, no mass effect was demonstrated.

There are many unanswered questions concerning the haemorrhagic risk of isolated DVAs and the physiological basis and anatomic source of haemorrhage when it coexists with cavernomas. Some reports separately attribute DVA-associated haemorrhage to thrombosis or

stenosis of central venous drainage, leading to a transient increase in pressure within the draining veins and thereby increasing the potential of venous haemorrhage [9].

The unenhanced CT brain scan demonstrated diffuse subcortical calcification in the right parieto-occipital region indicating chronic venous congestion in the brain. Despite this, the infant remained clinically well. But deterioration in the form of focal melting brain syndrome was to be expected.

In a series of 29 pediatric patients with dural arteriovenous shunts by Lasjaunias et Al [1], the overall outcome scores remain unsatisfactory. The therapeutic goal is not to erase what looks abnormal, but to make sure the existing venous outlets remain patent and functional. It was hoped that partial or staged treatment in this infant would promote the potential remodeling capacity of the venous drainage to her advantage. The main concern was that this infant only had the DVA to drain her right cerebral hemisphere which may not have the same remodeling capacity as the other cerebral veins.

The coexistent multiple cavernomas further complicated matters with episodes of haemorrhage. The decision to maintain the infant on low molecular heparin to preserve venous flow and limit thrombotic phenomenon proved to be difficult.

The unusual combination of disorders in this infant presenting simultaneously could be totally fortuitous. Any possible link could be hypothesized by the following explanation:

1. The complex disease entity demonstrated in this infant (table 1) appears to be a spectrum of phenotypic expression of a single (yet early) disorder. The dural sinus malformation, facial venous malformation, DVA and cavernous malformation on the right side seem to be in a metameric distribution, but her posterior fossa is normal. Could it be that dural development is segmented to account for the normal posterior fossa? Or could this presentation be a spectrum of the same disease with a specific target?

2. The various disorders (face, DVA, cavernoma and sinus malformation) could testify for an event at a point in time involving several, non generation related targets. The targets involved were those that were vulnerable at that time and though the diseases were not anatomically related, they were timely damaged simultaneously.

The actual genesis of this complex cerebrofacial vascular syndrome in this infant is still uncertain, though the second hypothesis could be the more likely explanation.

Conclusions

This is the first reported case of DSM associated with DVA, multiple cavernomas and facial venous malformation. The DSM also demonstrated postnatal enlargement which normally occurs in intrauterine life. The development of venous hypertension attributed by the DSM and the DVA increased the risk of venous haemorrhage of the co-existing cavernous venous malformation.

The treatment strategy was to correct venous hypertension by partial embolisation of the dural shunts to allow potential cerebral venous remodeling and preserve the patent sinus. Overall outcome scores remain unsatisfactory and deterioration in the form of focal melting brain syndrome is to be expected.

This complex disease appears to be in a metameric distribution but it could also testify for an event at a point in time involving several targets (not anatomically related) but vulnerable at that time and timely damaged simultaneously.

The actual genesis is still uncertain until further clinical, neuroradiologic and pathophysiological information is obtained to clarify the issue.

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